

Self-Injurious Behavior

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Self-Injurious Behavior

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Review article

Self-Injurious Behavior

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Abstract

HUISMAN, S., P. Mulder, J. Kuijk, M. Kerstholt, A. van Eeghen, A. Leenders, I. van Balkom, C. Oliver, S. Piening, R. Hennekam. Self-Injurious Behavior. *NEUROSCI BIOBEHAV REV* X(X) XXX-XXX, 2016-

Self-injurious behavior (SIB) is a relatively common behavior in individuals with intellectual disabilities (ID). Severe SIB can be devastating and potentially life-threatening.

There is increasing attention for somatic substrates of behavior in genetic syndromes, and growing evidence of an association between pain and discomfort with SIB in people with ID and genetic syndromes.

In this review on SIB phenomenology in people with ID in general and in twelve genetic syndromes, we summarize different SIB characteristics across these etiologically distinct entities and identify influencing factors. We demonstrate that the prevalence of SIB in several well-known genetic intellectual disability syndromes is noticeably higher than in individuals with ID in general, and that characteristics such as age of onset and topographies differ widely across syndromes. Each syndrome is caused by a mutation in a different gene, and this allows detection of several pathways that lead to SIB. Studying these with the behavioral consequences as specific aim will be an important step toward targeted early interventions and prevention.

Keywords:

Self-injurious behavior, intellectual disability, genes, genetic syndromes, Angelman Syndrome, Cornelia de Lange Syndrome, Cri du Chat Syndrome, Down Syndrome, fragile X

Syndrome, Lesch-Nyhan Syndrome, Lowe syndrome, Prader-Willi Syndrome, Rett Syndrome, Smith-Magenis Syndrome, Tuberous Sclerosis Syndrome, Williams-Beuren Syndrome, somatic factors, pain, interdisciplinary study.

Introduction

Self-injurious behavior (SIB) is a relatively common behavior in individuals with intellectual disabilities (ID). Severe SIB can be devastating and potentially life-threatening (Fig.1), and is associated with compromised mental health in parents and caregivers, high service needs and excessive health care costs (Wulffaert et al., 2009) (Hassiotis et al., 2008) (Ruddick et al., 2015) (Felce et al., 1998).

While there is abundant scientific interest in SIB in behavioral sciences, only limited attention is paid to SIB in medical sciences, despite increasing attention for somatic substrates of other behavior and evidence of an association between pain and discomfort with SIB (Brunner et al., 1993, Tint et al., 1994, Kishino et al., 1997, Breau et al., 2003, Symons et al., 2009, Symons, 2011, Diaz-Stransky and Tierney, 2012, Rosell and Siever, 2015, Wink et al., 2015). Studying specific genetic syndromes, with different molecular or metabolic etiologies, may show different characteristics of SIB depending on etiology, allowing various pathways leading to SIB to be discovered.

In this review we highlight twelve genetic syndromes in which sufficient phenomenological data are available: Angelman Syndrome (AS), Cornelia de Lange Syndrome (CdLS), Cri du Chat Syndrome (CdCS), Down Syndrome (DS), fragile X Syndrome (fraX), Lesch-Nyhan Syndrome (LNS), Lowe syndrome (LS), Prader-Willi Syndrome (PWS), Rett Syndrome (Rett), Smith-Magenis Syndrome (SMS), Tuberous Sclerosis Syndrome (TSC), and Williams-Beuren Syndrome (WBS).

The paper is based on a review of the literature concerning SIB studies with detailed analysis of phenomenology. Information regarding review methods and individual studies can be found in the Supplemental Materials.

Fig. 1: Individual with Cornelia de Lange Syndrome at 7, 21 and 38 years of age. Self-injurious behavior started before 7 years of age and deteriorated during puberty and adolescence. Hitting and head banging resulted in permanent sensory loss due to bilateral blindness, bilateral ear deformations and hearing impairments.

Conceptualization and definition

The term SIB was introduced by Tate and Baroff in 1966, to replace earlier labels such as masochism, auto-aggression, self-aggression and self-destructive behavior. Tate and Baroff stated that the term SIB did not imply an attempt to destroy, nor did it suggest aggression. It simply meant behavior that produces physical injury to the individual's own body (Tate and Baroff, 1966).

Subsequently, numerous authors have used variations of this definition (Table I). The main elements in definitions were: self-initiated; directed towards the body; involves specific forms and body parts; contains repetition; can be chronometrically or chronographically quantified (frequency, duration, intensity); and its effects or extent of tissue damage can be classified. Disqualifiers are intent of suicide or sexual arousal. Hence, we propose to define SIB as non-accidental behavior resulting in demonstrable, self-inflicted physical injury, without intent of suicide or sexual arousal. Typically the behavior is repetitive and persistent.

SIB prevalence

In this review twelve genetic syndromes are highlighted in which sufficient data were available: Angelman Syndrome (AS), Cornelia de Lange Syndrome (CdLS), Cri du Chat

Syndrome (CdCS), Down Syndrome (DS), fragile X Syndrome (fraX), Lesch-Nyhan Syndrome (LNS), Lowe syndrome (LS), Prader-Willi Syndrome (PWS), Rett Syndrome (Rett), Smith-Magenis Syndrome (SMS), Tuberous Sclerosis Syndrome (TSC), and Williams-Beuren Syndrome (WBS).

The paper is based on a review of the literature concerning SIB studies with detailed analysis of phenomenology. Detailed information regarding review methods and individual studies can be found in the Supplemental Materials.

SIB prevalence rates within and across genetic syndromes heavily depend on the methodology employed, i.e. definition, recruitment, sample characteristics and etiological diagnoses of ID. In populations of people with ID of unknown etiology, not stratified for age or levels of functioning, the prevalence of SIB in a non-residential care setting is ~30%, irrespective of age and level of cognitive functioning versus 41% in a residential care setting (Lowe et al., 2007)(Harris, 1993, Limdqvist, 2013)(Oliver et al., 2012)(Taylor et al., 2011)(Saloviita, 2000). If autism spectrum disorders (ASD) are present the prevalence rises markedly, varying from 42% to 70% (Buono et al., 2010, Eden et al., 2014). In a number of specific syndromes prevalence figures can differ strikingly from prevalence rates in individuals with ID in general, and can be very high (Fig. 2a). Highest prevalence rates have been reported in LNS, SMS, PWS, CdCS and CdLS.

Fig 2: Phenomenology of SIB in selected genetic syndromes, in ID of unknown origin and those with ID of unknown origin and ASD: a. Prevalence.

SIB has also been reported in a number of other genetic syndromes such as chromosome imbalances and non-genetically determined entities including teratogenic entities such as rubella encephalopathy and Fetal Alcohol Syndrome (May et al., 1981, Pace et al., 1986, Prasher et al., 1995, Engelen et al., 1999, Hoch et al., 2013, Plaisancie et al., 2014). The divergent prevalence figures for the twelve selected genetic syndromes and other entities advocate to organize early intervention, assessment and treatment strategies that are syndrome sensitive.

SIB phenomenology

Age of onset

Generally SIB starts in early childhood: 50% of individuals showed SIB before 3 years of age, 70% before 7 years of age up to 90% before 10 years of age; percentage rates in larger genetic syndrome studies vary from 12% < 1 year, 63% < 4 and 93% <11 years of age in fraX, versus 72% < 7 years of age in DS, and 73-91% < 7 years of age in PWS (Wigren and Heimann, 2001, Buono et al., 2005, Buono et al., 2010, Symons et al., 2010). Wide variation exists as in individual cases age of onset may occur in the first year of life, or SIB may first become manifest in adulthood. Case series in individuals with ID of unknown origin and in individuals with ASD demonstrated similar rates, but in the different syndromes median ages of onset vary widely (Fig. 2b). Rates may be prone to selection bias and can be overestimations, especially in small case series. Conversely, age of onset, may be underestimated when outcomes like physical damage is a criterion, and identifying SIB in childhood is difficult as behavior like self-hitting and even head banging may sometimes be judged as age appropriate behavior and is seen in typical development.

Fig 2: Phenomenology of SIB in selected genetic syndromes, in ID of unknown origin and those with ID of unknown origin and ASD: b. Age of onset.

Course of SIB

Figures about the course of SIB are sparse. SIB has been reported as persistent in 62% over 2 years, in 71% over 7 years and in 84% over 18 years (Taylor et al., 2011)(Emerson et al., 2001)(Cooper et al., 2009)(Oliver and Richards, 2015a). Long-term persistence of SIB is more likely with early onset and head directed self-injury (Emerson et al., 2001). SIB present at 20 years of age or older has an 84% chance to be chronic (Taylor et al., 2011). The relative risk of SIB increases until 30-40 years of age and starts to decrease after the age of 50 (Davies and Oliver, 2013).

Severity

Severity of SIB is determined by a combination of characteristics: chronography (frequency, duration), topography (form, localization, number), and physical damage. Both in research and patient care quantifying severity comprehensively is informative for comparisons of studies.

1. Chronography

Chronographic parameters (frequency, duration, intensity) can objectively quantify clinical severity and the effectiveness of interventions. These are parameters typically more accurately presented in case reports but less so in studies of larger cohorts. Therefore only limited data are available.

In general ID population studies SIB occurs occasionally in 27% and frequently in 14% (Saloviita, 2000); and every 30 min in 18%, hourly in 11%, daily in 43%, weekly in 19%, monthly in 8% and yearly in 1%, respectively (Saloviita, 2000) (Griffin et al., 1987). In various syndromes this can be markedly different: for instance in LNS SIB is typically almost continuously present and may even occur during sleep, without changing over time.

2. Topography

There are many forms of SIB involving divergent body sites, which are referred to as topographies. The most common forms are pulling (hair or nails), scratching, hitting, banging and biting. When hitting oneself a part of the body is typically used as the 'instrument' to hit, but objects can be used as well. Other common forms are inserting in orifice, picking, grinding, poking (eyes or ears), pinching and ruminating. Less common forms are mouthing, pica, crushing, snapping (neck), excessive drinking and air swallowing, sucking and choking (Fig. 2c). These less common forms can also occur without the other characteristics of a SIB and this behavior should not always be considered SIB. The most commonly involved body parts are the head, the hands and fingers. SIB remains confined to a single body part in 28%-46% of individuals, but involvement of several body parts frequently occurs, particularly in CdLS and SMS (Hall et al., 2001, Taylor et al., 2011, Petty et al., 2014).

Fig 2: Phenomenology of SIB in selected genetic syndromes, in ID of unknown origin and those with ID of unknown origin and ASD: c. Topographies. Some topographies have been indicated as being very characteristic for syndromes: this is based on the literature review and personal experiences but could not be determined statistically due to small numbers.

3. Physical damage

Physical damage as result of SIB is one of the main reasons for serious concerns of the caregivers, and for medical consultation. The physician needs not only to qualify but also to objectively quantify clinical severity, both to indicate immediate medical interventions and to judge the effectiveness of interventions. Although medical evaluation of physical damage is obligatory, very limited data have been reported on specific physical damage due to SIB. In seven of the twelve selected genetic syndromes in this review no data on severity of physical damage are provided. Detailed information on physical consequences in the remaining five syndromes has been best described in case reports. The Challenging Behavior Interview provides a four point Likert scale item on physical injury and the Self-Injury Trauma scale scores physical consequences in a subjective and time-consuming way, but otherwise there is no instrument available to the physician for scoring physical damage (Iwata et al., 1990, Oliver et al., 2003). Hence, we have designed a relatively simple scoring system for the physician defining the degree of severity based on the physical consequences of SIB and providing guidance on the need for further medical measures:

- 1- (relatively) mild: indicating non-permanent, minor tissue damage such as scratches, abrasion, bruises, temporary reddening of the skin, teeth marks;
- 2- moderate: indicating non-permanent, marked tissue damage or function loss, such as deep fissures, fractures, large scars and ulcerations;
- 3- severe: indicating permanent tissue loss, loss of sensory function (deafness; blindness), loss of neurological function (brain damage) and life-threatening consequences.

Since there are only small series that report information on tissue damage and function loss, it proved not to be possible to present results across genetic syndromes in a figure. We can only state reliably based on available literature and personal extensive experiences that

severe tissue damage is commonly prominent in LNS and CdLS. The frequent use of constraints in both syndromes is an indirect indication of the SIB severity.

Factors influencing SIB

A main issue in managing SIB is to assess and define factors predisposing, evoking and maintaining SIB as they indicate potential intervention strategies. These factors may be personal (gender; age), somatic (including genetic, neurobiological and medical conditions) and behavioral (including operant learning) in nature. Physicians in charge of SIB patients have a role that is complementary to that of the behavioral specialist, to evaluate health and functioning, paying specific attention to (painful) physical conditions which may lead to SIB. Behavioral specialists have a key role in assessing cognitive, adaptive and communicative abilities of SIB patients, in evaluating psychopathology that might be related to SIB and in performing a functional behavioral assessment.

1. Somatic

Somatic influencing factors of SIB in genetic syndromes (Fig. 2d) are subdivided in: genetic mutation, morphology, neurology, senses, gastro-intestinal, oral/dental, sleep, sensory and general health.

Larger numbers of different somatic influencing factors are reported in studies in individuals with ID of unknown origin, and also in CdLS and fraX. Senses (visual and hearing impairments) and other sensory (pain and tactile sense) problems are reported most across the various entities. However, most publications on SIB fail to report on even basic physical examinations by physicians who might evaluate potential physical causes of SIB, such as constipation, gastro-esophageal reflux disease (GERD)/esophagitis, intestinal obstruction,

dental problems, urinary tract infection, otitis media, sinusitis, presence of a foreign body, or fracture. Information on visual and hearing abilities is needed due to their high prevalence in individuals with ID, and their influence on adaptive and communicative abilities.

Furthermore, SIB is known to occur very frequently in several specific genetic syndromes and to occur in most of these genetic entities more frequently than in a population with ID of unknown origin (Fig. 2a). In the present analyses of studies on SIB the number of individuals in whom no or only partly genetic and metabolic diagnostic studies have been performed was remarkably high, which hampers optimal use of the knowledge of behavior in such entities, including the somatic substrates of behavior (Table II).

Fig 2: Phenomenology of SIB in selected genetic syndromes, in ID of unknown origin and those with ID of unknown origin and ASD: d. Influencing Factors.

2. Behavioral

Behavioral factors influencing SIB are presented in fig. 2d and for the overview divided in 2 categories: developmental (i.e. cognitive, communicative and adaptive abilities) and behavioral (intrapersonal and interpersonal). Intrapersonal characteristics include: stereotypy, repetitive, compulsive, impulsive behaviors, hyperactivity, distractibility, anxiety, and mood. Interpersonal behavioral characteristics encompass social contact and environmental dimensions such as adult attention, ignoring, demand avoidance, solitude, change of routine, changes to environment, institutionalization. ASD and aggression are presented separately. However, ASD as a distinctive etiology is problematic because ASD can be difficult to classify in these entities and symptoms of adaptive impairments and repetitive and stereotyped behaviors are also clustered within developmental and behavioral factors.

These characteristics influence behavior dysregulation and operant learning, and hence possible leads for behavioral interventions.

Discussion

SIB can be a devastating problem. It is devastating for the individuals who harm themselves, and who may experience a significant physical and psychological distress due to their SIB. It is devastating for the parents who see progressive damage to the one they love and who feel they cannot offer the protection they want to offer as a parent. It is devastating to caregivers who may feel ineffective and can experience this as a failure of their care.

Patients with SIB visit their general practitioners, pediatricians, child psychiatrists and pediatric neurologists, and later in life their internists, psychiatrists and even surgeons.

Managing SIB can be a huge challenge for physicians.

SIB is common in individuals with intellectual disability, often starts in early childhood and can aggravate into a destructive and persistent problem if interdisciplinary assessment and interventions are not applied. Accurate and detailed information on SIB characteristics such as chronography, topography and resulting physical damage may offer clues for the physician to the causes of pain or distress, either in the past or present, which may play a crucial role in the development and maintenance of SIB.

There is need for careful interdisciplinary evaluation of every patient who shows SIB. SIB should not be seen as a diagnosis, but as a symptom of an underlying problem. The physician must be alert to symptoms of underlying pain and discomfort as medical conditions causing or prolonging SIB may go unrecognized, undiagnosed and untreated, specifically due to the impaired communications skills in patients with ID. A comprehensive medical evaluation is

particularly valuable as frequent medical conditions like constipation, GERD, otitis media, dental problems, presence of a foreign body, or fracture, have excellent treatment options. Until now several theoretical models for SIB have been proposed, varying from neurobiological (including genetic and neurochemical), medical (pain and discomfort) and behavioral or operant learning models. There is growing evidence for an integrated biological and behavioral model in which genotypic-phenotypic characteristics and operant learning principles are complementary and lead to effective interventions (Oliver and Richards, 2015b) (Minshawi et al., 2015). Understanding the interactions between all these influencing factors needs longitudinal studies in which phenotyping the SIB characteristics and personal (developmental and behavioral) characteristics, the physical signs and symptoms of patients demonstrating SIB, and genotyping the same individuals, will be essential.

This review demonstrates that the prevalence of SIB in several well-known genetic ID syndromes is noticeably higher than in individuals with ID in general and that characteristics such as age of onset and topographies differ widely across syndromes, each caused by a different gene with a different action when mutated. Pathogenetic mechanisms behind these differences remain to be elucidated. It may be many different pathways can cause SIB. One may also hypothesize that these genes may have more than one action, one causing the syndrome and another causing SIB. Studying these multifunctional, 'moonlighting' proteins may show a common pathway to SIB (Jeffery, 2015). Comprehensive phenotype – genotype studies and functional analyses will be an important step towards targeted early interventions and effective prevention.

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We thank the family of the individual shown in Figure 1 for allowing us to show the pictures of their son.

Legends for Tables and Figures

Table I: Main Definitions for Self-Injurious Behavior Used in Literature.

Table II: Summary of a Literature Review of SIB in Genetic Syndromes.

Fig. 1: Individual with Cornelia de Lange syndrome at 7, 21 and 38 years of age. *Self-injurious behavior started before 7 years of age and deteriorated during puberty and adolescence. Hitting and head banging resulted in permanent sensory loss due to bilateral blindness, bilateral ear deformations and hearing impairments.*

Fig 2: Phenomenology of SIB in selected genetic syndromes, in ID of unknown origin and those with ID of unknown origin and ASD:

- a. Prevalence.
- b. Age of onset.
- c. Topographies. Some topographies have been indicated as being very characteristic for syndromes: this is based on the literature review and personal experiences but could not be determined statistically due to small numbers.
- d. Influencing Factors.

Subscripts for Fig. 2d:

*Behavioral: i.e. stereotypy, repetitive, compulsive, impulsive behavior, hyperactivity, distractibility, anxiety, prolonged distress, nervousness, mood, affect, tantrums, disturbing interpersonal behaviors)

****Social:** social contact, adult attention, ignoring, demand avoidance, automatic reinforcement, thwarting, boredom, solitude, being teased, frustration, change of routine, changes environment, length of institutionalization

Supplemental Materials

Appendix I Methods

Search strategies

Structured data extraction form

Appendix II Results

Figure S1 Study flow diagram

Table S1 Summary data of all included studies

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